An EcoRI polymorphism associated with a human genomic clone from band 11p13


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SOURCE AND DESCRIPTION OF PROBE: The probe (pS6H2.4) is a single copy 2.4 kb HindIII fragment subcloned from a lambda bacteriophage clone into pUC18. The bacteriophage clone is from a genomic library constructed from a Chinese hamster/human somatic cell hybrid containing only the short arm of human chromosome 11. Human DNA clones were identified by hybridization with a human Alu repeat sequence.

POLYMORPHISM: EcoRI (GAATTC) (Boehringer-Mannheim) detects a two allele polymorphism with a band at 6.7 kb or 3.6 kb.

ALLELE FREQUENCY: Ascertained in 18 unrelated individuals.

- 6.7 kb allele: 0.83
- 3.6 kb allele: 0.17

NOT POLYMORPHIC FOR: MspI, TaqI, HindIII, or PvuII with DNA from nine unrelated Caucasians.

CHROMOSOMAL LOCALIZATION: The probe has been localized to 11p13 by hybridization with a series of Chinese hamster/human somatic cell hybrids. It maps distal to the breakpoint of a translocation [t(4;11)(q22;p13)] segregating in a familial aniridia case. It maps within the region deleted in the somatic cell hybrid C2-1 [del(11)(p13p11)].

MENDELIAN INHERITANCE: Co-dominant segregation observed in three families (27 individuals).

PROBE AVAILABILITY: Generally available.


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